GENE**Tests**

Clinical Test Registration Form

Complete one Clinical Test Registration Form for each clinical test performed by the laboratory in-house.

Laboratory Director*			**Date***
Lab / Institution*			
Person completing this form*			
Phone*	**Fax**	**Email***	
GeneTests Disease Name (Search GeneTests by name or gene symbol to retrieve disease name)			**OMIM #**
OMIM Disease Name (optional)			

Molecular Genetic Testing (terms followed by ‡ are defined on page 3)

Gene symbol (one gene/form) **Chromosomal locus** **OMIM #**

Methods used to detect an unknown sequence variation within a region of DNA

Sequence analysis
- ☐ Sequence analysis of the entire coding region‡
- ☐ Sequence analysis of select exons‡ (list exons: required)

Mutation scanning
- ☐ Mutation scanning of the entire coding region‡
- ☐ Mutation scanning of select exons‡ (list exons: required)

Methods used to detect known (previously described) mutation(s)

Deletion/duplication analysis‡ (to detect copy number variation at one locus)
- ☐ PubMed ID or OMIM Allelic Variant Reference Number provided in "Comment" section verifying deletions/duplications cause this disease.
  Otherwise, the following statement will be included with your listing:
- ☐ No deletions or duplications involving _____ as causative of _____ have been reported. (Note: By definition, deletion/duplication analysis identifies rearrangements that are not identifiable by sequence analysis of genomic DNA.)

Targeted mutation analysis‡
- ☐ Nucleotide repeat expansion
- ☐ Single mutation or panel of mutations (does not include family-specific mutation analysis as such testing is not listed in the GeneTests Laboratory Directory)

Please list mutation(s), preferably according to the HGVS standard based on an explicit reference sequence (e.g. NG_005895.1:g.4323A>G):

Additional Methods

- ☐ Linkage analysis
- ☐ Methylation analysis
- ☐ Microsatellite instability testing (MSI)
- ☐ Uniparental disomy (UPD) study

Additional Molecular Genetic Testing Services

- ☐ Carrier testing‡ (Autosomal recessive or X-linked disorders only)
  - ☐ Carrier testing restricted to at-risk biologic family members only
  - ☐ Carrier testing restricted to members of specific ethnic group(s) only
  - ☐ Carrier testing restricted to at-risk biologic family members and members of specific ethnic group(s) only
- ☐ Prenatal diagnosis

Biochemical Genetic Testing

☐ Analyte ☐ Protein analysis ☐ Immunohistochemistry
☐ Enzyme assay ☐ Protein expression

Additional Biochemical Genetic Testing Services

☐ Carrier testing‡ (Autosomal recessive or X-linked disorders only)
- ☐ Carrier testing restricted to at-risk biologic family members only
- ☐ Carrier testing restricted to members of specific ethnic group(s) only
- ☐ Carrier testing restricted to at-risk biologic family members and members of specific ethnic group(s) only

☐ Prenatal diagnosis

Specialized Cytogenetic Testing

☐ Chromosome breakage studies
☐ FISH-interphase
☐ FISH-metaphase
☐ Multicolor FISH (M-FISH)/Spectral Karyotyping™ (SKY)™
☐ Sister chromatid exchange

Additional Specialized Cytogenetic Testing Services

☐ Carrier testing‡ (Autosomal recessive or X-linked disorders only)
- ☐ Carrier testing restricted to at-risk biologic family members only
- ☐ Carrier testing restricted to members of specific ethnic group(s) only
- ☐ Carrier testing restricted to at-risk biologic family members and members of specific ethnic group(s) only

☐ Prenatal diagnosis

Optional - Comments: Use this space to suggest information (not otherwise included on this form) to post for public display in the comment line of this disease listing; for example: description of tiered approach to testing, unusual sample requirements (e.g., muscle). This space may also be used for questions or comments for the GeneTests Staff and/or to provide PubMed ID or OMIM Allelic Variant Reference Numbers relating to deletion/duplication analysis. Please do not include sensitivity, sample requirements, turnaround time, shipping instructions, or costs.

Optional – Test-specific laboratory URL (if other than main lab URL):

Optional – Test-specific contact person[+] (will replace general lab contact[s] in test listing):

Contact email (required)
Contact phone (required)

*Required

[+]If this person is not registered in GeneTests: go to www.genetests.org; log in (Administrative Use); click “View, Add, or Edit Laboratory Information” for this laboratory; add person under Personnel. Then complete and submit this form.

‡Indicates term that is defined on page 3

E-mail, fax, or mail to GeneTests
E-mail (as attachment):gtlabs@u.washington.edu
Fax: 206-221-4679

Mailing address: GeneTests
9725 Third Ave NE, Suite 602
Seattle, WA 98115-0371

Methods Used To Detect an Unknown Sequence Variation within a Region of DNA

Sequence analysis of the entire coding region	(Synonyms: gene sequencing, sequence analysis, sequencing) The process by which the nucleotide sequence is determined for the entire coding region of a gene
Mutation scanning of the entire coding region	(Synonyms: scanning, mutation screening) A 2-step process by which the entire coding region of a gene is first analyzed via one of a variety of methods (such as CSGE, DGGE, SSCP, DHPLC or TGCE) to identify sequence alterations. These methods do not identify the specific nucleotide change(s) and must be followed by further analysis (usually sequencing) to identify the specific sequence alteration
Sequence analysis of select exons	The process by which specific exons are sequenced to identify sequence variations; used to expedite analysis when certain exons are likely to contain the disease-causing mutation(s)
Mutation scanning of select exons	A 2-step process by which specific exons of a gene are first analyzed via one of a variety of methods (such as CSGE, DGGE, SSCP, DHPLC or TGCE) to identify sequence alternations. These methods do not identify the specific nucleotide change(s) and must be followed by further analysis (usually sequencing) to identify the specific sequence alteration

Methods Used To Detect a Known (Previously Described) Mutation

Family-specific mutation analysis	Testing for the specific disease-causing mutation(s) previously identified in a family member. Note: Family-specific mutation analysis is different from targeted mutation analysis and is not listed in the GeneTests Laboratory Directory
Targeted mutation analysis	(Synonym: allele-specific mutation analysis) Testing for either (1) a nucleotide repeat expansion (e.g., the trinucleotide repeat expansion associated with Huntington disease), or (2) one or more specific mutations (e.g., Glu6Val for sickle cell anemia, a panel of mutations for cystic fibrosis). Deletion/duplication analysis and family-specific mutation analysis are excluded from this definition.
Deletion/duplication analysis	(Synonym: copy number analysis) A process to detect deletions/duplications of an entire exon, multiple exons, or the whole gene that typically are not identifiable by sequence analysis of genomic DNA. Many methods may be used, including quantitative PCR, real-time PCR, multiplex ligation dependent probe amplification (MLPA), and array CGH. (Note: Gene-targeted array CGH may be listed by disease in the GeneTests Laboratory Directory, but multi-disorder (multi-locus) screening array CGH can only be listed as a Miscellaneous test.)

Other Definitions

Array genomic hybridization	(Synonyms: AGH, aGH, array GH, copy number variation (CNV) analysis. Related terms: array CGH, aCGH, array comparative genomic hybridization, chromosomal microarray analysis, oligonucleotide array comparative hybridization, oligo aCGH, oligonucleotide microarray analysis) A method of examining multiple loci simultaneously to identify genetic imbalance caused by the gain or loss of chromosomal material ranging in size from a whole chromosome to about 80-500 kb.
Carrier testing	(Synonyms: carrier detection, heterozygote testing) Testing used to identify usually asymptomatic individuals who have a gene mutation for an autosomal recessive or X-linked disorder
Immuno-histochemistry	Testing to detect the presence of specific proteins in cells or tissues by means of a specific antigen/antibody reaction tagged with a visible label
Protein expression	Testing to examine the expression of a mutation in a recombinant protein to confirm its pathogenicity